

Table SI. Diagnostic Criteria for BCNS

Major criteria

1. BCC prior to 20 years old or excessive numbers of BCCs
2. OKCs of the jaw prior to 20 years of age
3. Palmar or plantar pits
4. Lamellar calcification of the falx cerebri
5. Medulloblastoma, typically desmoplastic
6. First-degree relative with BCNS
7. *PTCH1* or *SUFU* gene mutation in normal tissue*

8. Minor criteria

1. Rib anomalies
2. Other specific skeletal malformations and radiologic changes^a
3. Macrocephaly
4. Cleft lip or palate
5. Cardiac or ovarian fibroma
6. Lymphomesenteric cysts
7. Ocular abnormalities^b

a: vertebral anomalies, kyphoscoliosis, short fourth metacarpals, postaxial polydactyly

b: strabismus, hypertelorism, congenital cataract, glaucoma, coloboma

BCC: basal cell carcinoma; OKC: odontogenic keratocyst; BCNS: basal cell nevus syndrome.

A diagnosis of BCNS was made when one fulfilled either two major criteria, or one major and two minor criteria.

*Must be accompanied by at least one other major criterion for diagnosis.

Table SII. Fulfillment of diagnostic criteria in the 18 patients with BCNS

Classic characteristics	All patients (18) n(%)	Mutation detected (6) n (%)
50% or greater		
Basal cell carcinoma	16 (88.9)	4 (66.7)
Biopsy prior to 20 y/o	2 (11.1)	0 (0.0)
Excessive numbers of BCCs (>2 BCC)	13 (72.2)	3 (50.0)
Odontogenic keratocysts (OKC)	15 (83.3)	6 (100)
OKC diagnosed prior to 20 y/o	6 (33.3)	3 (50.0)
Lamellar calcification of the falx cerebri	13 (72.2)	6 (100)
Palmar or plantar pits	13 (72.2)	6 (100)
15-49%		
Other specific skeletal malformations	9 (50.0)	4 (66.7)
Ocular anomalies	8 (44.4)	6 (100)
Macrocephaly	7 (38.9)	5 (83.3)
First degree relative affected#	6 (33.3)	3 (50.0)
Rib anomalies	5 (27.8)	4 (66.7)
Medulloblastoma, typically desmoplastic	3 (16.7)	1 (16.7)
14% or less		
Cleft lip or palate	2 (11.1)	1 (16.7)
Cardiac or ovarian fibroma	2 (11.1)	0 (0.0)
Lymphomesenteric cysts	0 (0.0)	0 (0.0)

* $P < .05$; ** $P < .01$; *** $P < .001$

three of the four patients belong to the same family

y/o: years old; BCC: basal cell carcinoma.

Table SIII. Comparison of findings of Basal Cell Nevus Syndrome in different studies

Characteristics	Kimonis et al USA (1997)	Rehefeldt-Erne et al Switzerland (2016)	Endo et al Japan (2012)	Our study Taiwan (2024)
Sex M:F	1:1.2	1:0.76	1.1:1	1:1
Mean age at first BCC (years)	21.4	24	37.4	37.5
Basal cell carcinoma	71/90 (80)	23/28 (82.11)	56/148 (37.8)	16/18 (88.9)
OKC of the jaw	78/105 (74)	25/28 (89.3)	126/146 (86.3)	15/18 (83.3)
Lamellar Calcification of the falx cerebri	53/82 (65)	11/13 (84.6)	78/98 (79.6)	13/18 (72.2)
Palmar or plantar pitting	89/102 (87)	17/28 (60.7)	86/143 (60.1)	13/18 (72.2)
Medulloblastoma	4/105 (4)	0	4/120 (3.3)	3/18 (16.7)
First degree relative with BCNS	NA [◆]	15/30 (50)	62/128 (48.4)	6/18 (33.3)

◆NA, not assessed; BCC: basal cell carcinoma; OKC: odontogenic keratocyst; BCNS: basal cell nevus syndrome.